



ANKH gene

ANKH inorganic pyrophosphate transport regulator

Normal Function

The *ANKH* gene provides instructions for making a protein that is present in bone. This protein transports a molecule called pyrophosphate out of cells to the intricate network of proteins that forms in the spaces between cells (extracellular matrix). Pyrophosphate helps regulate bone formation by preventing mineralization, the process by which minerals such as calcium and phosphorus are deposited in developing bones. The *ANKH* protein may have other, unknown functions.

Health Conditions Related to Genetic Changes

craniometaphyseal dysplasia

At least seven mutations in the *ANKH* gene have been found to cause craniometaphyseal dysplasia. Some mutations change a single protein building block (amino acid) in the *ANKH* protein, whereas others insert or delete an amino acid in the *ANKH* protein. These mutations most likely decrease the protein's ability to transport pyrophosphate out of cells. Reduced levels of pyrophosphate increase bone mineralization, which may contribute to the bone overgrowth seen in craniometaphyseal dysplasia.

other disorders

About five mutations in the *ANKH* gene have been found to cause a rare hereditary form of calcium pyrophosphate dihydrate deposition disease (CPPDD). CPPDD, also called chondrocalcinosis or pseudogout, is characterized by the accumulation of calcium pyrophosphate dihydrate crystals in the cartilage of joints. The buildup of these crystals weakens cartilage and causes it to break down more easily. The crystals may cause pain and inflammation in the joints. Most cases of CPPDD occur in people older than 40, are not inherited, and have an unknown cause.

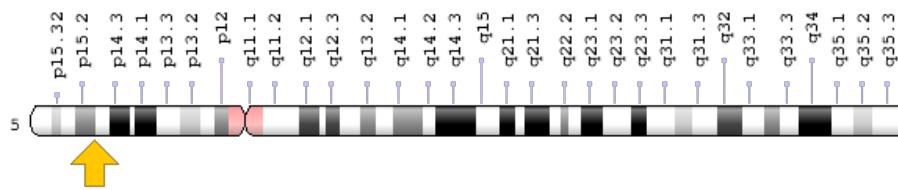
Mutations in the *ANKH* gene have been shown to cause CPPDD in a few families. In these families, one altered copy of the *ANKH* gene in each cell is sufficient to cause the condition. Individuals with familial CPPDD caused by *ANKH* mutations typically form crystal deposits within multiple joints during early adulthood. Researchers believe that these mutations lead to elevated pyrophosphate levels in the extracellular matrix. High levels of pyrophosphate result in excessive formation of calcium pyrophosphate dihydrate crystals within joints.

Studies suggest that certain variations (polymorphisms) in the *ANKH* gene are associated with the normal difference in bone size and shape among individuals. These polymorphisms probably result in slight changes in the activity of the *ANKH* protein, affecting the levels of pyrophosphate in the extracellular matrix.

Chromosomal Location

Cytogenetic Location: 5p15.2, which is the short (p) arm of chromosome 5 at position 15.2

Molecular Location: base pairs 14,704,800 to 14,871,778 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ANK
- ANKH_HUMAN
- ankylosis, progressive homolog (mouse)
- FLJ27166
- HANK
- MANK
- progressive ankylosis protein

Additional Information & Resources

Educational Resources

- Merck Manual Professional Version: Calcium Pyrophosphate Dihydrate Deposition Disease
<http://www.merckmanuals.com/professional/musculoskeletal-and-connective-tissue-disorders/crystal-induced-arthritides/calcium-pyrophosphate-dihydrate-crystal-deposition-disease>
- Molecular Biology of the Cell (fourth edition, 2002): Bone Is Continually Remodeled by the Cells Within It
<https://www.ncbi.nlm.nih.gov/books/NBK26889/#A4187>

GeneReviews

- Craniometaphyseal Dysplasia, Autosomal Dominant
<https://www.ncbi.nlm.nih.gov/books/NBK1461>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ANKH%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- ANK, MOUSE, HOMOLOG OF
<http://omim.org/entry/605145>
- CHONDROCALCINOSIS 2
<http://omim.org/entry/118600>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_ANKH.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ANKH%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=15492

- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/56172>
- UniProt
<http://www.uniprot.org/uniprot/Q9HCJ1>

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